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FULL PAPER

Description of the MRI and ultrasound imaging features of giant epidermal cysts

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Objectives: Guidelines suggest that lesions over 5 cm in dimension should be referred to a specialist sarcoma centre due to the possibility of malignancy. Few epidermal cysts (ECs) reach or exceed this size and are termed giant ECs (GECs). The purpose of this study is to report on a large series of GECs.

Methods: Retrospective review of histologically proven GECs over an 8-year period. Patient demographics with MRI and ultrasound (US) appearances were evaluated.

Results: A total of 14 cases were included with eight males and six females. Mean age was 51 years. 11 lesions were oval and three bi-lobed in shape, while 12 demonstrated dermal apposition. All were hyperintense on water-sensitive sequences and isointense to slightly hypointense on T1W imaging. Internal clefts were seen in 13 cases and 11 demonstrated chemical shift artefact (CSA) on MRI. On US, 12 showed well-defined linear hypoechoic clefts, with 66.6% having dis-organised compared with 33.3% peripherally located clefts. One

'pseudo testis' pattern and one showing irregular striped echogenicity termed novel 'pseudo muscle' appearance. No cases demonstrated internal vascularity on Doppler US.

Conclusions: MRI signal findings of GECs are often characteristic with hyperintensity on water-sensitive sequences, dermal apposition, CSA and internal clefts while US features of disorganised or clumped hypoechoic clefts and absence of neovascularity were commonly seen. Recognition of combinations of both US and MRI features of GECs should reduce the requirement for pre-excisional needle biopsy to confirm the diagnosis.

Advances in knowledge: 1. Identification of common imaging features of GECs should avoid unnecessary pre-excisional biopsy despite their large size in the appropriate MDT setting.

2. A novel 'pseudo-muscle' appearance is described on MRI and US.

INTRODUCTION

Epidermal cysts (EC) are common subcutaneous lesions that rarely cause diagnostic concern, and in the majority of cases are not formally imaged but excised by general surgeons, plastic surgeons and increasingly general practitioners. They are usually asymptomatic masses measuring <4 cm in diameter with a predilection for hair bearing areas of the face, neck and trunk. Although they can occur anywhere on the body, <10% involve the extremities.¹ Pathologically, they are filled with keratin debris surrounded by a wall of stratified squamous epithelium² and are thought to occur either due to traumatic transplantation of epidermal cells into the dermis, or possibly being congenital in nature with aberrant embryogenesis.³ Lesions > 5 cm in maximal diameter have been termed 'giant epidermal cysts' (GECs), with only a few case reports in the literature describing their clinical and imaging features.⁴⁻¹¹

With regard to soft tissue tumours in general, clinical features suggestive of malignancy include large masses and those exhibiting recent growth, with any soft tissue lesion >5 cm in maximum diameter quoted as having a 20% likelihood of being malignant.¹² The National Institute for Health and Care Excellence (NICE) guidelines state that if a patient presents with a mass lesion >5 cm, they should be promptly referred to a specialist centre.¹³ This is supported by the European Society of Skeletal Radiologists (ESSR) approved guidelines for diagnostic imaging in soft tissue tumours in adults, which also suggests MRI for investigation of any lesion >5 cm in size.¹⁴ The purpose of our study is to provide a descriptive pictorial review of the imaging appearances of pathologically proven GECs and also to report a novel 'pseudomuscle' imaging appearance on both MRI and ultrasound (US).

METHODS

The study was approved by the local Research and Innovation Centre of the Institute of Orthopaedics with no requirement for informed patient consent. A retrospective search of the pathology database for all patients who had a confirmed histological diagnosis of EC during the period of 1 June 2009 to 2 June 2017 was undertaken, in the setting of a tertiary musculoskeletal oncology referral centre. A total of 56 patients had a diagnosis of EC, of which 14 had a single orthogonal length of >5 cm, and would therefore be defined as GECs. These formed the final study group. All 14 cases had histopathological diagnosis from percutaneous biopsy, with only one patient undergoing surgical resection at our institution.

Patient demographics including age and sex together with lesion location, size and imaging features were recorded. The MRI studies demonstrated a wide variety of protocols due to referral from different institutions. However, all studies were considered to be of diagnostic adequacy at multidisciplinary team (MDT) meetings. T1W turbo spin echo (T1W TSE) images were available in all 14 cases, T2W fast spin echo (T2W FSE) sequences in 8, and short tau inversion recovery (STIR) sequences in 11 cases. Post-contrast studies were obtained in eight patients.

MRI studies were evaluated for lesion location, size, shape, dermal apposition and signal intensity characteristics. T1W TSE, STIR and T2W FSE SI were qualitatively compared as being hyperintense, isointense or hypointense compared to skeletal muscle. The presence of chemical shift artefact (CSA), at the periphery of the lesion on non-fat saturated short TE sequences was also recorded. Internal debris or clefts and their signal characteristics were also assessed, being defined as intralesional curvilinear or branch-like foci. The enhancement pattern was recorded.

US was performed with a variety of probes, mostly with a 15 MHz linear array probe, but images were also obtained with a 5 MHz curvilinear array probe due to the larger size of some lesions. 'Static' US images were evaluated in consensus with respect to lesion location relative to the dermis, shape and imaging characteristics including internal clefts and debris, and

for neovascularity on Doppler. The clefts or debris were defined as intralesional curvilinear or branch-like hypoechoic foci.

RESULTS

There were eight males and six females, with a mean age of 51 years (range 20–78 years). Lesion locations included the thigh ($n = 4$), shoulder ($n = 3$), arm ($n = 2$), flank, buttock, forefoot, lower leg and supraclavicular regions ($n = 1$). Maximal lesion dimensions ranged from 50 to 90 mm, with a mean of 64 mm. Eleven of 14 (78.6%) GECs were oval in shape (Figures 1–4), with 3 (21.4%) being bi-lobed in appearance (Figures 5 and 6). The bi-lobed lesions were in the forefoot, arm and lower leg.

Magnetic resonance imaging

On T1W TSE images, eight lesions (57.1%) were isointense (Figures 1a and 4a) while six (42.9%) were isointense-to-mild hypointense compared to skeletal muscle (Figure 6). On T2W FSE images, 8 of 8 (100%) and on STIR 11 of 11 (100%) lesions were hyperintense (Figure 1c). 13 cases (92.9%) demonstrated well-defined, linear internal clefts or debris which appeared hyperintense on T1W TSE images in 12 of 13 cases (92.3%), while in 7 of 8 cases (87.5%), the clefts were hypointense on T2W FSE images, and in 11 of 11 cases (100%), they had hypointense STIR signal intensity (Figure 1a). One (7.1%) lesion demonstrated alternating iso- and hyperintense bands extending over the entire diameter of the lesion (Figure 4a and b).

12 (85.7%) showed apposition to or bulging into the overlying dermis on MRI (Figures 1, 4a, b and 5a and b), which was corroborated on static US images (Figures 2–5c). Of interest, both lesions with no definite dermal apposition were bi-lobed in appearance (Figure 6). Six of eight (75%) GECs demonstrated well-defined, thin peripheral post-contrast enhancement (Figure 5b), with a further 2 (25%) demonstrating no discernible enhancement. CSA was identified at the lesion margin in 11 of 14 cases (78.6%) on non-fat saturated sequences (Figure 1b).

Ultrasound

Ultrasound confirmed the MRI appearances of the shape of the lesion and its relationship with the dermis, with 11 of 14 (78.6%)

Figure 1. A 40-year-old male with a subcutaneous GEC in the proximal right thigh. (a) Coronal T1W TSE (TR 790; TE 20) and (b) axial T2FS FSE (TR 4000; TE 96) MR images demonstrating a large oval isointense T1W TSE and hyperintense STIR signal intensity lesion (arrows) with multiple internal hyperintense T1 and isointense T2FS clefts. Dermal apposition (white arrow) and chemical shift artefact (black arrows) are well demonstrated.

Figure 2. A 65-year-old female with a subcutaneous GEC in the thigh. Longitudinal US image demonstrating a large oval lesion with internal anechoic foci (black arrows) and echogenic reflectors (white arrows).

demonstrating an oval appearance (Figures 2–4c) and 3 (21.4%) a bi-lobed appearance (Figure 5c). 12 of 14 (85.7%) demonstrated an association with the dermis (Figures 2–5c), and 12 of 14 (85.7%) contained filiform anechoic areas with two separate appearances identified. Eight of 12 (66.6%) showed irregular disorganised linear or branch-like filiform anechoic clefts (Figure 2), while 4

of 12 (33.3%) demonstrated well-defined ‘clumps’ or ‘aggregates’ of filiform anechoic areas at the periphery of the lesion (Figure 3). One (7.1%) demonstrated a ‘pseudo testis’ appearance with small hypo- and hyperechoic foci (Figure 5c), the US features resembling those of a normal testis. One lesion demonstrated alternating

Figure 3. A 78-year-old male with a subcutaneous GEC in the buttock. Longitudinal US image demonstrating a large oval lesion with internal anechoic foci clumped at the periphery of the lesion (white arrows). The dermal apposition is demonstrated.

Figure 4. A 78-year-old male with a subcutaneous GEC in the left shoulder. (a) Coronal T1W TSE (TR 827;TE 12), (b) fat saturated post-contrast T1W TSE MR images (TR 561;TE 10) and (c) longitudinal US demonstrating a large lesion with alternating intrinsic linear strands resulting in a 'pseudomuscle' appearance. There is loss of fat plane between the lesion and dermis (white arrows) consistent with dermal apposition.

hypo- and hyperechoic bands extending over the entire diameter of the lesion (Figure 4c).

A summary of the imaging features is presented in Table 1.

DISCUSSION

ECs are also known as epidermoid cysts, epidermal inclusion cysts, sebaceous cysts and infundibular cysts. Clinically, they occur in the dermis or subcutaneous tissues as mobile lumps with a central punctum.¹⁵ They are uni-locular without separation and usually grow very slowly over years or decades. Histologically, they demonstrate stratified squamous epithelium with an admixture of internal components¹⁶ including variable amounts of keratinous debris with intervening fragments of packed keratin lamellae, foreign body reactions, sporadically distributed cholesterol or sebaceous foci and possible dystrophic calcifications. The compact keratin lamella has been reported to form a concentric ring pattern,³ which gives rise to the typical concentric imaging appearances described in the literature, particularly in the testes.¹⁷

As anticipated, dermal apposition was demonstrated in 85.7% of our cases, which is similar to the previously reported figure of 88.1% from 42 cases of varying sizes assessed with US.³ Interestingly, one of the lesions in our study that did not demonstrate dermal apposition showed a very clear bi-lobar appearance in the lower leg with peripherally sited internal clefts representing a multilocular giant EC (MGEC), which has been previously described in the literature.^{18,19} Other previously reported features of MGEC include a bi-lobed appearance, with common occurrence in elderly males on the trunk and soles of the feet at sites of thickened skin.¹⁹ The other bi-lobed lesion was present in the intermetatarsal space of the foot with the anatomical location of extrinsic pressure from the metatarsals likely to account for the morphology. However, most lesions in our study demonstrated an oval shape (78.6%), as expected.

The signal intensity characteristics of GECs in the current study are consistent with the appearances described in case reports^{6,11} and several other studies on smaller ECs, which ranged from 40 to 56% isointense signal and 44–60% slightly hypointense signal compared to skeletal muscle on T1W TSE sequences.^{20,21} Hong

Figure 5. A 44-year-old male with a subcutaneous GEC in the fourth intermetatarsal space of the left forefoot. (a) Coronal pre- and (b) post-contrast fat-saturated T1W TSE (TR 500; TE 19) MR images demonstrating a bi-lobed lesion with peripheral, thin rim enhancement. (c) US image of the same lesion demonstrating 'pseudotestis' pattern with echogenic reflectors (white arrows) and anechoic areas (black arrows) and dermal apposition (white arrowheads).

et al²² reported more varied T1W signal characteristics with hypointense (21%), isointense (50%) and hyperintense (29%) signal. The variety in signal characteristics is thought to be due to differences in composition of the cyst during various stages of maturation.

ECs are also described in both the radiology and histopathology literature as containing serpentine, mixed signal intensity internal debris.²² Internal debris has been previously documented as being present in 44–100% of cases.^{20,21} Internal debris was confidently identified on MRI in 93% of cases in the current study, with the only negative case demonstrating a previously

Figure 6. A 63-year-old female with a subcutaneous multilocular giant epidermal cyst (MGEC) in the lower leg. Coronal T1W TSE (TR 409.91; TE10) MR image demonstrating a bi-lobed lesion deep to the dermis (black arrows) with intrinsic hyperintense T1 filiform structures (white arrows).

undescribed 'pseudomuscle' appearance. There is debate as to the aetiology of internal debris. In one study demonstrating irregular low signal intensity areas within the lesion on both T1W and T2W imaging, the debris was confirmed as representing aggregates of keratinous material on pathological examination.²³ Huang³ postulated that the intralesional bright echogenic foci on US may be secondary to cholesterol, sebaceous foci or calcification, whereas the anechoic areas may be ascribed to scattered fragments of packed keratin lamellae. This is in contrast to other authors,^{17,21,24} who suggested that the floating linear echogenic reflections were created by layered and aggregated keratin debris, with the dark clefts occurring secondary to cyst-like spaces and areas lacking keratin. In the current study, the clefts demonstrated heterogeneous signal characteristics presumably representing a mixture of proteinaceous clefts, lipid aggregates and cholesterol.

Chemical shift artefact (CSA) occurs due to mis-registration from the different resonance frequencies of fat and water, and results in artefactual linear hypointensity at fat-fluid interfaces. The literature is divided as to whether this occurs in the frequency²⁵ or phase-encoding direction²⁶ with artefacts in spin echo (SE) imaging due to frequency encoding direction (as

Table 1. Summary of GECs demographic and imaging features

Patient and lesion demographics			
		Sex	Male 8
			Female 6
		Age	Mean 51 years (range 20–75 years)
		Shape and location	Oval 11 (thigh $n = 4$, shoulder $n = 3$, arm $n = 2$, flank, buttock, forefoot, lower leg, supraclavicular $n = 1$)
			Bi-lobed 3 (forefoot, lower leg, arm)
		Size	Mean 64 mm (range 50–90 mm)
MRI		Deep dermal involvement	12 of 14 (85.7%)
	Signal T1	isointense	8 of 14 (57.1%)
		hypointense	6 of 14 (42.9%)
	STIR	hyperintense	14 of 14 (100%)
	Intrinsic clefts		13 of 14 (92.9%)
		Hyperintense T1 clefts	7 of 8 (87.5%)
		Hypointense STIR clefts	11 of 11 (100%)
	Post contrast enhancement	Thin peripheral	6 of 8 (75%)
		Nil	2 of 8 (25%)
	Chemical shift artefact		11 of 14 (78.6%)
US		Deep dermal involvement	12 of 14 (85.7%)
	Intrinsic morphology	Linear alternating pattern	1 of 14 (7.1%)
		Pseudotestis	1 of 14 (7.1%)
		Filiform anechoic foci	12 of 14 (85.7%)
	Location intrinsic filiform foci	Disorganised	8 of 12 (66.6%)
		Peripheral clumping	4 of 12 (33.3%)

probably demonstrated in this study due to TSE sequences protocols used in our institution) and in gradient echo (GE) imaging in the phase-encoding direction since the GE sequence lacks the 180° degree refocusing pulse. Nearly 80% of lesions in this study demonstrated CSA at the lesion margins, suggesting this could be a useful accessory diagnostic imaging feature. Given that these lesions generally contain compact keratin, it is not altogether easy to explain this artefact, but it may be due to intrinsic cyst-like spaces that have been previously described.^{17,21,24} However, recent literature has expressed caution with regard to the usefulness of this CSA in the characterisation of superficial soft tissue masses, concluding that it is not an infrequent imaging finding which is of no value in differentiating benign from malignant lesions.²⁷

The US features of ECs are well-documented, with hyporeflexive dermally apposed lesions described in the literature.^{1,3,21} A 'pseudotestis' appearance has been documented to occur in

62% of cases of ECs,³ which had a mean dimension of 0.5 cm and range up to 1 cm.³ Only one patient in our series had this finding. A more common US appearance was that of disorganised (57%) or peripherally located (27%) clumps or aggregates of internal anechoic foci. Therefore, this may represent changes due to maturation and enlargement of the cyst and its contents infrequently seen in smaller lesions³ but the authors recognise this is conjecture in view of the lack of radiologic-pathological correlation. In addition, 93% of GECs in our series demonstrated no internal neovascularity which may be a further helpful diagnostic feature, considering that most solid benign and malignant soft tissue lesions demonstrate internal hypervascularity on Doppler US.^{28,29}

One lesion showed linear alternating signal intensity bands on MRI that the authors have described as a novel 'pseudomuscle' appearance with corresponding alternating echogenic linear strands across the entire diameter of the lesion on US (Figure 4).

We hypothesise that this appearance may be secondary to alternating layers of dense laminated keratin orientated in a linear rather than concentric ring pattern, an imaging appearance that is widely recognised in epidermoid cysts of the testis,^{3,21} although the authors acknowledge that no formal radiologic-pathological correlation was possible as formal surgical excision was not undertaken at our institution. We are not aware of this appearance being described in the literature previously, and therefore it should be added to the potential imaging features of GECs.

Malignant degeneration of ECs is recognised but uncommon, having been reported in approximately 2% of over 3000 ECs.³⁰ More recently, squamous cell carcinoma (SCC) within an EC wall has been described, with literature review identifying 16 further case reports of such malignant degeneration.³¹ Interestingly, 9 of the 14 (64.3%) cases were GECs by size criteria.³¹ We did not have a case of malignant degeneration in our series, and therefore cannot comment on the possible imaging features but the most recent case report clearly demonstrated a 'solid' nodule associated with the cyst wall which should raise suspicion for malignant degeneration.³¹ There is also a report of intramuscular extension of an epidermoid cyst that was thought to be secondary to chronic rupture,¹¹ but none of our cases demonstrated this finding. A further study concluded that most ruptured EC have septa, thick irregular rim enhancement with associated fuzzy enhancement in the surrounding subcutaneous tissues that may simulate a mass of infectious or neoplastic origin.²²

The differential diagnosis of cystic lesions in the dermis and subcutaneous layer includes neurogenic lesions, myxomatous tumours and lymphatic malformations or any lesions that may undergo cystic degeneration or haemorrhage. If there are any red flag clinical features such as a Tinel's sign, then consideration should also be given to a neurogenic lesion.

There are several limitations of our study that include the relatively small sample size, although this series represents the largest study of GECs. The wide variety of MRI sequences due to lesions being imaged prior to referral meant that no standard imaging protocol was available. The retrospective nature of the study did not allow for MRI/US-pathology correlation to confirm potential hypotheses for the imaging characteristics. In particular, radiologic-pathologic correlation was not available for the lesion with the novel 'pseudomuscle' imaging appearance, while confirmation of the aetiology of internal debris or potential cystic-spaces to explain the CSA was not possible. Advanced MRI techniques such as diffusion-weighted imaging (DWI) were not included in the study due to the wide variety of referral cases. DWI has been postulated to aid in soft tissue tumour diagnosis by assessing intrinsic cellularity,³² with specific description in the literature of subcutaneous ECs returning significantly lower attenuation diffusion coefficient (ADC) values compared with those of intracranial epidermoid cysts.³³ Some authors advocate the utilisation of DWI in imaging protocols for assessment of soft tissue masses^{32,34} although this is not currently included routinely at our institution.

CONCLUSION

The MRI signal characteristics of GECs are often characteristic, with 100% demonstrating hyperintensity on water-sensitive sequences. Dermal apposition, CSA, internal clefts and no or thin rim enhancement on MRI, with absence of intrinsic neovascularity on Doppler US are all very common features. The US features are varied, but anechoic foci within the lesions in either a disorganised or peripherally clumped arrangement are commonly seen. This combination of MRI and US findings in these large lesions in the appropriate clinical and MDT setting should allow a confident imaging diagnosis and avoid unnecessary pre-excisional percutaneous needle biopsy. A novel alternating linear 'pseudomuscle' appearance is described that should be added to the potential imaging features of ECs.

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